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CASE REPORT

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Disabling Head Tremor in a Patient with DYT1 Mutation

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Dystonic head tremor is known to be a feature in some patients with DYT1 mutation. However, isolated tremor of the head without relevant cervical dystonia has rarely been described. We report here a patient with the three-bp GAG deletion in the DYT1 gene (904_906delGAG) who had severe head tremor in the frame of a generalized limb dystonia.

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Key Words: DYT 1 gene, Dystonia, Tremor.

The GAG deletion in the DYT1 gene usually causes a typical form of primary torsion dystonia with early onset in a limb, rapid generalization, and sparing of cranial-cervical muscles,[1,2] but atypical phenotypes have often been reported, including late age at onset, limited spread of dystonia and involvement of neck and facial muscles.[3-6]

We describe here a patient with DYT1 mutation who had severe head tremor in the absence of relevant cervical dystonia.

Case Report

52-year-old man visited to our clinic complaining of disabling head tremor. Initially, he developed dystonic twisting his right foot especially while walking at mid 30s. Dystonia spread progressively over 10 years to involve the left foot. At the same time, he noted head tremor, which worsened progressively, leading to severe problems in eating and drinking since 5 years ago. He also developed excessive eye blinking and forceful eye closure 3 years ago. He denied family history of any similar disorder. On examination, the patient had a coarse, slow (3-4 Hz), complex head tremor, with a combination of "yes-yes" and "no-no" oscillations, worsening on turning his head to right or left. In the upper limbs, there was a combined postural and action tremor with similar frequency, interfering with writing. On holding the arms outstretched, mild dystonic posturing was also noted. There was no apparent neck deviation. In neutral position, blepharospasm and only mild right-convex scoliosis was noted. He got some relief by holding her forehead lightly, resting his head against the back of a chair, or lying down. Head tremor was aggravated by emotional stress and disappeared while he was sleeping. Dystonic posturing, inversion and plantar flexion, of the both feet was appeared at rest and prominent while walking. His eye movements were normal. Detailed cognitive function tests revealed no abnormalities. There was no motor weakness or sensory deficits. Deep tendon reflexes were symmetrically normoactive. Plantar reflexes were flexor bilaterally. Routine laboratory tests were all normal as well as measurements of serum ceruloplasmin, CK, and lactate levels. Electroencephalography and brain MRI were unremarkable. Various medical treatment including trihexyphenidyl, clonazepam, and propranolol were ineffective. Alcohol also failed to provide any benefit. Genetic testing confirmed the three-bp GAG deletion in the DYT1 gene (904_906delGAG).

Discussion

Although the spectrum of dystonia produced by the DYT1 GAG deletion is broad, in-

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cluding both childhood and adult onset and variable progression of signs from focal to widespread involvement, the typical phenotype usually is early-onset dystonia developing before the age of 26 years in a limb, particularly the leg, and rapidly generalizes within 5 years from onset.[1,2] Selective or pronounced cranio-cervical involvement is said to be atypical in DYT1 dystonia.[1-3] Onset of dystonia in the facial or neck muscles appears to be a much rarer feature. In the vast majority of cranio-cervical involvement, dystonia begins in a limb, and subsequently spread to the cranial-cervical region in the frame of a generalized phenotype.[3]

Tremor in DYT1 dystonia is usually dystonic tremor or rarely isolated postural hand tremor.[4,5,7] Head tremor accompanying cervical dystonia is known to be a feature in some patients.[4,6] However, disabling head tremor in the absence of relevant cervical dystonia has rarely been described in DYT1 dystonia. Although co-occurrence of essential tremor was a diagnostic consideration, some of the clinical features shown by this patient would be quite unusual for such a diagnosis. He used maneuvers to attenuate the head tremor, which were similar to "sensory tricks" observed in patients with cervical dystonia. There was also striking change in his tremor on his head position, which is typical of dystonia.[8] Isolated head tremor associated with these features may be initial manifestation of cervical dystonia.[8] In these cases, neck deviation did not become apparent for many years, and eventually developed obvious cervical dystonia. Therefore, long-term clinical follow-up will confirm the nature of his head tremor.

We describe here atypical clinical features in a patient with DYT1 mutation who had isolated disabling head tremor in the frame of a generalized limb dystonia.

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